CLAIMS

What is claimed is:

1. An isolated nucleic acid molecule comprising a nucleotide sequence selected from the group consisting of:

a) SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14 and 15; and

b) the complement of SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14 and 15.

2. An isolated nucleic acid molecule comprising an exon from a vertebrate gene wherein said exon is at least 1150 base pairs in length.

3. An isolated nucleic acid molecule according to Claim 2, wherein said gene is a human gene.

4. An isolated nucleic acid molecule according to Claim 2, wherein said gene is a *spastin* gene.

5. An isolated nucleic acid molecule consisting of a nucleotide sequence selected from the group consisting of:

a) SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14 and 15; and

b) the complement of SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14 and 15.

6. An isolated portion of a nucleic acid sequence selected from the group consisting of:

a) SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14 and 15; and

b) the complement of SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14 and 15,

wherein the portion is at least about 10 nucleotides in length.

7. A nucleic acid molecule comprising a nucleotide sequence which is at least about 60% identical to a nucleotide sequence selected from the group consisting of:

a) SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14 and 15; and

b) the complement of SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14 and 15.

8. A nucleic acid molecule which hybridizes under high stringency conditions to a nucleotide sequence selected from the group consisting of:

a) SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14 and 15; and

b) the complement of SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14 and 15.

9. A nucleic acid construct comprising the isolated nucleic acid molecule of Claim 1.

10. The nucleic acid construct of Claim 9 wherein the isolated nucleic acid molecule is operatively linked to a regulatory sequence.

11. A recombinant host cell comprising the isolated nucleic acid molecule of Claim 1.

12. The recombinant host cell of Claim 11 wherein the isolated nucleic acid is operatively linked to a regulatory sequence.

13. A method for preparing a polypeptide encoded by an isolated nucleic acid molecule, comprising culturing the recombinant host cell of Claim 12.

14. An isolated polypeptide encoded by an isolated nucleic acid molecule according to Claim 1.

15. An isolated polypeptide encoded by an isolated nucleic acid molecule according to Claim 5.

16. An antibody, or an antigen-binding fragment thereof, which selectively binds to the polypeptide encoded by an isolated nucleic acid molecule according to Claim 1, or to a portion of said polypeptide.

17. A method for assaying the presence of a nucleic acid molecule in a sample, comprising contacting said sample with a nucleotide sequence selected from the group consisting of:

a) SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14, 15, 17-66, 72 and 73;

b) the complement of SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14, 15, 17-66, 72 and 73;

c) a portion of any one of SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14, 15, 17-66, 72 and 73 which is at least 10 nucleotides in length; and

d) a portion of the complement of any one of SEQ ID NOS: 1, 3, 7, 9, 11, 12, 13, 14, 15, 17-66, 72 and 73 which is at least 10 nucleotides in length under conditions appropriate for selective hybridization.

18. A method for assaying the presence of a polypeptide encoded by an isolated nucleic acid molecule according to Claim 1 in a sample, comprising contacting said sample with an antibody which specifically binds to the encoded polypeptide.

19. An isolated polypeptide comprising an amino acid sequence selected from the group consisting of SEQ ID NOS: 2, 4, 8, 10, 16 and 67-69.

20. An isolated polypeptide comprising an amino acid sequence having greater than 75 % identity to an amino acid sequence selected from the group consisting of SEQ ID NOS: 2, 4, 8, 10, 16 and 67-69.

21. An antibody which specifically binds to the polypeptide of Claim 19.

22. An antibody which specifically binds to the polypeptide of Claim 20.

23. An isolated nucleic acid molecule consisting of a nucleotide sequence selected from the group consisting of:

a) SEQ ID NOS: 21-66; and

b) the complement of SEQ ID NOS: 21-66.

24. A method of diagnosing or aiding in the diagnosis of neurodegenerative disease in an individual comprising

a) obtaining a nucleic acid sample from the individual; and

b) determining the nucleotide present at nucleotide position 5254 of SEQ ID NO: 1,

wherein presence of a thymine at said position is indicative of increased likelihood of neurodegenerative disease in the individual as compared with an individual having a cytosine at said position.

25. The method of Claim 24, wherein said neurodegenerative disease comprises one or more symptoms selected from the group consisting of: reduced sensory nerve conduction, reduced motor nerve velocity, hypermyelination of retinal nerve fibers, atrophy of upper cerebellar vermis, absence of Purkinje cells and abnormal neuronal lipid storage.

26. The method of Claim 24, wherein the nucleic acid sample is obtained from a tissue selected from the group consisting of: brain tissue, CNS, lung, fetal lung, testis, lymphocytes, adipose, fibroblasts, skeletal muscle, pancreas, uterus, kidney, tonsil, embryo and isolated cells thereof.

27. The method of Claim 26, wherein said brain tissue is selected from the group consisting of cerebral cortex, granular cell layer of the cerebellum and hippocampus.

28. The method of Claim 24, wherein the neurodegenerative disease is an early onset neurodegenerative disease.

~~29.~~ A method of diagnosing or aiding in the diagnosis of neurodegenerative disease in an individual comprising

a) obtaining a nucleic acid sample from the individual; and

b) determining whether there is a deletion of a thymine at nucleotide position 6594 of SEQ ID NO: 1,

wherein deletion of a thymine at said position is indicative of increased likelihood of neurodegenerative disease in the individual as compared with an individual who does not have a deletion at said position.

30. The method of Claim 29, wherein said neurodegenerative disease comprises one or more symptoms selected from the group consisting of: reduced sensory nerve conduction, reduced motor nerve velocity, hypermyelination of retinal nerve fibers, atrophy of upper cerebellar vermis, absence of Purkinje cells and abnormal neuronal lipid storage.

31. The method of Claim 29, wherein the nucleic acid sample is obtained from a tissue selected from the group consisting of: brain tissue, CNS, lung, fetal lung, testis, lymphocytes, adipose, fibroblasts, skeletal muscle, pancreas, uterus, kidney, tonsil, embryo and isolated cells thereof.

32. The method of Claim 31, wherein said brain tissue is selected from the group consisting of cerebral cortex, granular cell layer of the cerebellum and hippocampus.

33. The method of Claim 29, wherein the neurodegenerative disease is an early onset neurodegenerative disease.

~~34.~~ A method of treating a neurodegenerative disorder associated with the presence of a thymine at nucleotide position 5254 of SEQ ID NO: 1 in an individual, comprising administering to the individual an agent selected from the group consisting of:

a) a polypeptide encoded by SEQ ID NO: 2 or an active portion thereof;

b) a nucleic acid molecule which encodes SEQ ID NO: 2 or an active portion of SEQ ID NO: 2; and

c) an agonist of SEQ ID NO: 2.

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~~35.~~ A method of treating a neurodegenerative disorder associated with a deletion at nucleotide position 6594 of SEQ ID NO: 1 in an individual, comprising administering to the individual an agent selected from the group consisting of:

a) a polypeptide encoded by SEQ ID NO: 2 or an active portion thereof;

b) a nucleic acid molecule which encodes SEQ ID NO: 2 or an active portion of SEQ ID NO: 2; and

c) an agonist of SEQ ID NO: 2.

~~36.~~ A method of diagnosing or aiding in the diagnosis of neurodegenerative disease associated with the presence of a thymine at nucleotide position 5254 of SEQ ID NO: 1 in an individual, comprising:

a) obtaining a sample comprising a Spastin polypeptide from the individual;

b) determining the size of the Spastin polypeptide,

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wherein if the Spastin polypeptide is significantly shorter than SEQ ID NO: 2 it is indicative of neurodegenerative disease.

37. A method according to Claim 36, wherein the Spastin polypeptide is significantly shorter than SEQ ID NO: 2 if the Spastin polypeptide comprises less than about 75% of the amino acids of SEQ ID NO: 2.

~~38.~~ A method of diagnosing or aiding in the diagnosis of neurodegenerative disease associated with the presence of a deletion at nucleotide position 6594 of SEQ ID NO: 1 in an individual, comprising:

a) obtaining a sample comprising a Spastin polypeptide from the individual;

b) determining the size of the Spastin polypeptide,

wherein if the Spastin polypeptide is significantly shorter than SEQ ID NO: 2 it is indicative of neurodegenerative disease.

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39. A method according to Claim 36, wherein the Spastin polypeptide is significantly shorter than SEQ ID NO: 2 if the Spastin polypeptide comprises less than about 75% of the amino acids of SEQ ID NO: 2.